very long-chain acyl-CoA dehydrogenase deficiency

Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency is a condition that prevents the body from converting certain fats to energy, particularly during periods without food (fasting).

Signs and symptoms of VLCAD deficiency typically appear during infancy or early childhood and can include low blood sugar (hypoglycemia), lack of energy (lethargy), and muscle weakness. Affected individuals are also at risk for serious complications such as liver abnormalities and life-threatening heart problems. When symptoms begin in adolescence or adulthood, they tend to be milder and usually do not involve the heart.

Problems related to VLCAD deficiency can be triggered by periods of fasting, illness, and exercise. This disorder is sometimes mistaken for Reye syndrome, a severe disorder that may develop in children while they appear to be recovering from viral infections such as chicken pox or flu. Most cases of Reye syndrome are associated with the use of aspirin during these viral infections.

Frequency

VLCAD deficiency is estimated to affect 1 in 40,000 to 120,000 people.

Genetic Changes

Mutations in the *ACADVL* gene cause VLCAD deficiency. This gene provides instructions for making an enzyme called very long-chain acyl-CoA dehydrogenase, which is required to break down (metabolize) a group of fats called very long-chain fatty acids. These fatty acids are found in foods and the body's fat tissues. Fatty acids are a major source of energy for the heart and muscles. During periods of fasting, fatty acids are also an important energy source for the liver and other tissues.

Mutations in the *ACADVL* gene lead to a shortage (deficiency) of the VLCAD enzyme within cells. Without sufficient amounts of this enzyme, very long-chain fatty acids are not metabolized properly. As a result, these fats are not converted to energy, which can lead to the characteristic signs and symptoms of this disorder such as lethargy and hypoglycemia. Very long-chain fatty acids or partially metabolized fatty acids may also build up in tissues and damage the heart, liver, and muscles. This abnormal buildup causes the other signs and symptoms of VLCAD deficiency.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- ACADVL
- acyl-CoA dehydrogenase very long chain deficiency
- very long-chain acyl coenzyme A dehydrogenase deficiency
- very long-chain acyl-coenzyme A dehydrogenase deficiency
- VLCAD-C
- VLCAD deficiency
- VLCAD-H

Diagnosis & Management

These resources address the diagnosis or management of VLCAD deficiency:

- Baby's First Test
  http://www.babysfirsttest.org/newborn-screening/conditions/very-long-chain-acyl-coa-dehydrogenase-deficiency
- GeneReview: Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK6816
- Genetic Testing Registry: Very long chain acyl-CoA dehydrogenase deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342784/
- MedlinePlus Encyclopedia: Newborn Screening Tests
  https://medlineplus.gov/ency/article/007257.htm

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Newborn Screening Tests
  https://medlineplus.gov/ency/article/007257.htm
- Health Topic: Lipid Metabolism Disorders
  https://medlineplus.gov/lipidmetabolismdisorders.html
- Health Topic: Newborn Screening
  https://medlineplus.gov/newbornscreening.html

Genetic and Rare Diseases Information Center

- VLCAD deficiency
  https://rarediseases.info.nih.gov/diseases/5508/vlcad-deficiency

Educational Resources

- Disease InfoSearch: VLCAD deficiency
  http://www.diseaseinfosearch.org/VLCAD+deficiency/7410
- Genetics Education Materials for School Success (GEMSS)
  http://www.gemssforschools.org/conditions/vlcad/default
- Illinois Department of Public Health
  http://www.idph.state.il.us/HealthWellness/fs/mcad.htm
- MalaCards: very long-chain acyl-coenzyme a dehydrogenase deficiency
  http://www.malacards.org/card/very_long_chain_acyl_coenzyme_a_dehydrogenase_deficiency
- Medical Home Portal
  https://www.medicalhomeportal.org/newborn/vlcadd
- My46 Trait Profile
  https://www.my46.org/trait-document?trait=Very%20long-chain%20acyl-CoA%20dehydrogenase%20deficiency&type=profile
- New England Consortium of Metabolic Programs
  http://newenglandconsortium.org/for-families/other-metabolic-disorders/fatty-acid-oxidation-disorders/vlcadd/
- Orphanet: Very long chain acyl-CoA dehydrogenase deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=26793

- Screening, Technology, and Research in Genetics
  http://www.newbornscreening.info/Parents/fattyaciddisorders/VLCADD.html
- Virginia Department of Health
  http://www.vdh.virginia.gov/content/uploads/sites/33/2016/11/Parent-Fact-Sheet_VLCAD_English.pdf

Patient Support and Advocacy Resources

- Children Living with Inherited Metabolic Diseases (CLIMB)
  http://www.climb.org.uk/
- Children's Mitochondrial Disease Network (UK)
  http://www.cmdn.org.uk/
- FOD (Fatty Oxidation Disorders) Family Support Group
  http://www.fodsupport.org/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/very-long-chain-acyl-coa-dehydrogenase-deficiency-lcad/
- United Mitochondrial Disease Foundation
  http://www.umdf.org

GeneReviews

- Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK6816

Genetic Testing Registry

- Very long chain acyl-CoA dehydrogenase deficiency
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0342784/

ACT Sheets

- Elevated C14:1 +/- other long-chain acylcarnitines
  https://www.ncbi.nlm.nih.gov/books/NBK55827/bin/C14.pdf

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22very+long-chain+acyl-coenzyme+a+dehydrogenase+deficiency%22

Scientific articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28very+long-chain+acyl+coenzyme+a+dehydrogenase+deficiency%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D

OMIM

- ACYL-CoA DEHYDROGENASE, VERY LONG-CHAIN, DEFICIENCY OF
  http://omim.org/entry/201475

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